MATERNAL SERUM ALPHA-FETO PROTEIN

SCREENING TEST

A blood test used to discover certain birth defects during pregnancy

Glossary

Alpha-fetoprotein (AFP): a substance present in a woman's body during pregnancy. It is made by the fetus and is detectable as early as 29 days after conception. AFP passes from the amniotic fluid through the placenta into a pregnant woman's blood. The AFP level can be measured at specific times during pregnancy in the mother's blood or amniotic fluid to check for the presence or absence of certain birth defects of the developing baby.

Amniotic Fluid: the liquid surrounding the fetus.

Down Syndrome: a combination of birth defects and mental retardation resulting from extra genetic material.

Fetus: the unborn baby.

Maternal Serum: one part of a pregnant woman's blood.

Maternal Serum Alpha-fetoprotein (MSAFP) Test: a blood screening test used in early pregnancy primarily to detect neural tube defects in the fetus. It is also used as a screening test for Down Syndrome.

MSAFP Triple Test (also known as AFP Profile or AFP Plus): a new screening test that measures MSAFP as well as other substances in maternal blood. This test is thought to be a better screening test for Down Syndrome.

Neural Tube Defects: an opening in the neural tube, the structure that forms the brain and spinal cord during the fourth week of pregnancy (for example, Spina Bifida and Anencephaly).

Placenta: the flat, round organ attached to the wall of the uterus during pregnancy. This organ makes it possible for the growing baby to get oxygen and nutrients from its mother and assist in getting rid of waste products (also called the lifeline or afterbirth).

Screening Test: screening tests used during pregnancy identify women who may have a fetus with certain birth defects from those who probably do not.

A screening test does not prove that the fetus has a birth defect but it will indicate whether further testing may be necessary.

MSAFP ✓

Pregnant women now have a blood test available to them called the Maternal Serum Alpha-fetoprotein (MSAFP) Screening Test which is offered between the 16th and 18th weeks of pregnancy. This test measures the amount of a substance in maternal serum called alpha-fetoprotein (AFP). AFP is a substance that is produced by the fetus and is passed from the amniotic fluid through the placenta into the mother's blood circulation where it can be measured.

The MSAFP screening test may provide valuable information to pregnant women about their developing babies. About 950 out of 1,000 women will have a normal test. Elevated levels of MSAFP may suggest that a woman is carrying twins or that she may be further along in her pregnancy than she originally thought.

***However,* in approximately 2 out of every 1,000 women tested, an elevated MSAFP level may indicate a birth defect of the brain or spinal cord of the fetus, called a neural tube defect. About 95% of infants with neural tube defects are born to women with no prior family history of the condition.**

Other rare birth defects or pregnancy related conditions may also be identified by this screening test.*

Because the MSAFP test is a screening test, it can only suggest the presence or absence of an abnormality and indicate whether further testing might be considered.

If the MSAFP screening test shows an abnormally high amount of AFP in the blood, the test may be repeated; if the level remains elevated, further tests such as ultrasound, and, if necessary, amniocentesis can be used to look for the cause of the abnormal level.

As the development of new prenatal tests increases, pregnant women have many difficult choices and decisions to consider. It is recommended that individuals considering the MSAFP screening test read this brochure and discuss the details of the screening and testing process with both their partner and their health care provider.

***For more information regarding the use of this test in screening for Down Syndrome, see page 5.**

What birth defects can the MSAFP screening test detect?

This blood test can detect most open neural tube defects in the unborn baby.

Neural tube defects are defects in the tube that forms the brain and spinal cord during the fourth week of pregnancy. Normally, the neural tube closes completely. If, however, all or part of the neural tube fails to close, the fetus will have a neural tube defect.

In some instances, the opening of the tube may be left exposed; in others, this defect may be closed or covered by skin. Most closed neural tube defects are **not** detected by the MSAFP screening test.

Certain rare birth defects affecting the intestines or internal organs, or other conditions may also be discovered by this test.

What are some types of neural tube defects?

There are different forms of neural tube defects, varying widely in their severity. Two common types are anencephaly and spina bifida. Both occur as a result of the brain or spinal cord failing to develop properly before a child is born.

Anencephaly (an-en-SEF-a-lee): In this disorder, much of the brain and skull are abnormally formed. Anencephaly is such a severe disorder that a baby with this problem may be stillborn or usually dies shortly after birth, even with the best medical attention.

Spina Bifida (SPI-nah BIF-i-da): The degree of spinal malformation and nerve damage caused by this disorder varies.

In most instances, the spinal cord protrudes from the back. This may result in varying degrees of paralysis, loss of sensation in the lower limbs, and in bowel and bladder complications. Hydrocephalus ("water on the brain") can be a complication of spina bifida. In some instances, mental retardation may occur.

However, children with spina bifida may develop normally with little physical handicap and without mental retardation. Some individuals with spina bifida walk by themselves. Others walk

with braces, crutches or use wheelchairs. Treatment of children born with spina bifida has greatly improved in recent years. Children are usually treated in major medical centers which provide corrective surgery and other needed therapy.

With a "team" approach to health care (that includes parents, doctors, therapists, and educators), children born with spina bifida can reach their full potential and lead satisfying lives.

Are neural tube defects common?

In the United States, the majority of babies are born healthy! Only 1-2 in every 1,000 babies born will have a neural tube defect.

The specific cause of these defects is not known. They result from a combination of inherited and environmental factors.

Most babies with neural tube defects (about 95%) are born to women who have no "special risk" characteristics, such as having a known personal or family history of this disorder. Women who have already given birth to a child with neural tube defect, or parents who have a family history of this disorder do have a greater chance for having a baby with a neural tube defect. Genetic counseling can help a woman understand her chance of having a baby with a birth defect.

When should a pregnant woman have the MSAFP screening test?

If you choose to have the MSAFP screening test, it should be done 16-18 weeks after the first day of your last menstrual period. The timing of this test is extremely important for accurate results.

Unreliable results may be obtained if the blood sample is taken too early or too late during pregnancy. If the first test results are elevated, adequate time must remain in the pregnancy for further steps in the testing process.

To do the MSAFP screening test, a sample of blood is taken from the arm and is sent to a laboratory. The laboratory tests the blood sample to see how much AFP is present then sends the results to the health care provider.

The results of the MSAFP screening test are usually known within 1-2 weeks after the blood sample is obtained.

What if the MSAFP screening test results are normal?

About 950 out of every 1,000 pregnant women will have a normal MSAFP screening test. If normal levels of AFP are found, the test is considered normal and further testing is not indicated.

Women should understand that an MSAFP screening test in the normal range cannot guarantee a normal baby at birth. However, it can offer assurance that the baby is not likely to have an open neural tube defect.

What if the MSAFP level is elevated?

When the MSAFP level in the first blood sample is elevated, the results are not considered final. An elevated result of the MSAFP test does not necessarily mean a neural tube defect is present. Often it means that the pregnancy is further along than initially thought (AFP normally increases in the blood as pregnancy progresses) or that a woman is carrying twins.

Because this is a screening test, approximately 48-49 out of every 1,000 women may have elevated results even though they are carrying normal, healthy babies. *Only in 1 or 2 out of every 1,000 women will an unusually high MSAFP level indicate a neural tube defect.*

When the first sample is elevated, a new blood sample should be requested by the health care provider and the test will be repeated.

If the second MSAFP test is normal, further testing is usually not indicated.

If the second MSAFP test is elevated, other prenatal diagnostic tests, such as ultrasound and, if necessary, amniocentesis can be done to look for the cause of the elevated levels.

What if the MSAFP level is low?

Recent research indicates that low MSAFP values may be helpful in identifying pregnant women under 35 years of age who may have a higher likelihood of carrying a fetus with Down Syndrome.

It is possible for a woman in any age group to have an infant with Down Syndrome but the small chance increases as a woman gets older. The use of MSAFP as a screening test for Down Syndrome may help the population of women *under* 35 years of age who are not normally offered amniocentesis to diagnose this birth defect.

If the first MSAFP sample is low, the results are not considered final. Using a sophisticated chart, information such as the MSAFP result, maternal age, weight and week of pregnancy is considered to calculate a "risk figure" for Down Syndrome. This is more accurate than the risk figure usually calculated which is based on maternal age alone. Based on this new information, ultrasound and possibly amniocentesis may be offered by the health care provider. The ultrasound may reveal that the woman isn't as far along in the pregnancy as was originally thought, and that her MSAFP test result may actually be normal for her stage in pregnancy. If the ultrasound doesn't explain the low level, women will choose whether or not to have an amniocentesis to analyze the genetic make-up of the fetus and accurately determine if the fetus has Down Syndrome.

It is very important to remember that the MSAFP blood test does not diagnose Down Syndrome. It is a screening test used to identify some, but not all, women who may have a higher likelihood of carrying a fetus with the disorder. Amniocentesis is the procedure which can precisely diagnose this condition. If you want to learn more about your specific chances for having a baby with Down Syndrome, seek genetic counseling.

What is the MSAFP Triple Test (also known as AFP Profile or AFP Plus)?

This test is a new screening test that measures MSAFP as well as other substances in maternal blood. It is thought to be a better screening test for Down Syndrome. Research on this new test is currently underway. At this time, this test is not being used routinely and is not a replacement for the regular MSAFP screening test.

What is Down Syndrome?

Down Syndrome is a genetic condition which results from an extra #21 chromosome derived from either the sperm or the egg. Down Syndrome is also called Trisomy 21, and in the past, it was known as Mongolism. People with Down Syndrome have a characteristic physical appearance and mental retardation which can range from mild to severe. The condition may be complicated by congenital heart disease and/or respiratory problems. With early evaluation, education and other resources and supports, many individuals with Down Syndrome can live productive lives in community settings.

What are ultrasound and amniocentesis?

Ultrasound or sonography (son-NOG-rah-fee) is a procedure which uses sound waves to obtain a picture of the fetus. A special device (called a transducer) is moved across the mother's abdomen to view the fetus.

This picture will enable the doctor to tell whether the blood test was abnormal because of twins or because the week of pregnancy was estimated incorrectly. Ultrasound can also show the shape of the fetal skull and spine and may be able to detect certain neural tube defects as well as other birth defects.

If sonography does not give a reason for the abnormal MSAFP values, amniocentesis may be recommended by the doctor.

Ultrasound is generally considered to be safe for the mother and fetus although at present, the long-range effects are unknown.

Amniocentesis (am-nee-o-sen-TEE-sis) needs to be performed by an experienced physician, usually in an office or clinic setting.

This procedure is usually performed after the 16th week of pregnancy for prenatal genetic diagnosis. Ultrasound is used

simultaneously to help the doctor locate the position of the fetus and the placenta.

A needle is inserted through the abdomen into the woman's uterus and a small amount of amniotic fluid is withdrawn in a syringe. The amount of AFP in the amniotic fluid is then measured to test for neural tube defects. If the amniotic fluid AFP level is high and other possible causes have been ruled out, there is a substantial chance that the fetus has a neural tube defect. The combination of the MSAFP blood screening test, ultrasound, and amniocentesis can diagnose over 95% of all open neural tube defects.

Other tests can be done using the amniotic fluid to determine if genetic conditions (such as Down Syndrome) are either present or absent.

Amniocentesis is considered to be safe when performed by an experienced physician. However, possible complications that can develop from the procedure are bleeding, infection, miscarriage and/or injury to the fetus.

What if all tests are abnormal?

This is a difficult time for some women and their partners. Some women may be shocked and saddened, or may experience periods of fear, guilt, shame, anger, or denial. These are all normal feelings that are shared to some degree by all parents who learn their developing baby has a birth defect.

By gathering information and discussing facts and feelings with family members, health care providers, counselors, and other parents who have had a similar experience, an informed decision about the pregnancy can be made.

If the fetus is found to have a birth defect, after careful thought, a woman may choose to continue the pregnancy or to terminate the pregnancy. If a woman chooses to continue the pregnancy, the obstetrician can make special arrangements for delivery in a hospital or medical center with expertise in the care and surgery that is often required immediately after birth for newborns with birth defects.

Genetic Counseling

Genetic counseling, which is provided by health professionals, can help a family learn about their chances of having a baby with a birth defect. A genetic counselor can help individuals learn about available tests to check for the presence or absence of birth defects. Many individuals find it helpful to discuss these complex issues with a genetic counselor because it allows them to learn what options are available so that they can make more informed decisions.

Should you have the MSAFP screening test?

The MSAFP screening test is voluntary. Since many children born with birth defects are born to women without family histories of the disorder, this is a decision every pregnant woman now has to consider. In order to make an informed decision, both parents, if possible, should discuss this test with their health care provider.

If you choose to have the MSAFP screening test, you should understand in detail what each step in the screening and testing process means and what your options are. You should understand that it is possible that a birth defect may be missed and that there is a very slight chance that a healthy fetus may be incorrectly identified as having a birth defect.

For Your Notes:

After reading this brochure, it is advised that you discuss the MSAFP screening process with your health care provider—before asking for or consenting to have the MSAFP screening test. Consent can be withdrawn at any time in the screening or diagnostic process.

If you would like to learn more about the MSAFP screening test or the availability of genetic counseling or diagnostic services, please talk with your obstetrician, or you can write or call:

The Massachusetts Genetics Program
Division of Perinatal Health
Bureau of Parent, Child and Adolescent Health
Massachusetts Department of Public Health
150 Tremont Street, 2nd Floor
Boston, MA 02111
(617) 727-5121

Other Resources

Spina Bifida Association of America
1700 Rockville Pike, Suite 250
Rockville, MD 20852
(301) 770-7222
or
1-800-621-3141

Massachusetts Spina Bifida Association
24 Wentworth Road
Melrose, MA 02176
(617) 239-1919

National Down Syndrome Congress
1800 Dempster Street
Park Ridge, IL 60088
(312) 823-7550
1-800-232-6372

Massachusetts Down Syndrome Congress
124 Greenwood Lane
Waltham, MA 02154
(617) 742-4440

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